



NOTES

COLORECTAL POLYP CONDITIONS

GENERALLY, WHAT ARE THEY?

PATHOLOGY & CAUSES

- Colorectal polyps: **overgrowths** of epithelial cells **lining colon/rectum**
- Usually benign, can turn malignant

TYPES

Adenomatous polyps/colonic adenomas

- Gland-like polyps caused by tumor suppressor **gene mutation** in **adenomatous polyposis coli** (APC)
- Characterized by accelerated division of epithelial cells → epithelial dysplasia → polyp formation
- No malignant potential by itself**; requires mutations in other tumor suppressants (K-RAS, p53)
- Histologic classification
 - Tubular**: pedunculated polyp, protrudes out in lumen
 - Villous**: sessile, cauliflower-like appearance; more **often malignant**
 - Tubulovillous**: characteristics of tubular, villous polyps

Serrated polyps

- Saw-tooth** appearance microscopically
- Contain **methyated CpG islands** → silencing of DNA-repair genes, others → more mutations → malignancy
 - Small polyps (most common)**: AKA hyperplastic polyps; rarely malignant
 - Large polyps**: often flat, sessile, malignant

Hamartomatous polyps

- Mixture of tissues; disorganized mass containing tissue found at site of polyp
- Occur sporadically/in **genetically inherited conditions** (Juvenile polyposis, Peutz-Jeghers syndrome)

Inflammatory polyps

- Caused by **inflammatory bowel diseases**
 - Crohn's disease, ulcerative colitis
- Not malignant

CAUSES

- Genetic mutations
- Inflammatory conditions (e.g. Crohn's disease)

RISK FACTORS

- Family history
- Bowel wall injury (e.g. radiation exposure, smoking, inflammatory bowel disease)
- Risk increases with age

COMPLICATIONS

- Malignancy
 - Depends on degree of dysplasia, size of polyp

SIGNS & SYMPTOMS

- Often **asymptomatic**
- If ulcerating
 - Rectal bleeding, anemia symptoms (e.g. fatigue)
- If large
 - Obstruction → abdominal pain, constipation
- Malabsorption → diarrhea
- Some polyposis syndromes
 - Extracolonic symptoms

VISIT...

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DIAGNOSIS

DIAGNOSTIC IMAGING

CT scan, MRI

- Hyperdense outpouchings of colonic wall into lumen; detection of metastases

Endoscopy (colonoscopy) with biopsy

- Type of polyp, malignant potential (degree of dysplasia)

LAB RESULTS

- Iron-deficiency anemia → decreased red blood cell (RBC) count, low mean corpuscular volume (MCV) levels
- Iron-deficiency anemia → low ferritin, serum iron, transferrin saturation
- APC, RAS, etc. mutations
- Assess asymptomatic family members for risk

OTHER DIAGNOSTICS

Digital rectal examination

- Detection of distal rectal polyps; malignant polyp, hard, irregular; benign polyps, softer, pliable



Figure 29.2 Histological appearance of a villous adenoma, characterised by a surface composed of long villous projections.

TREATMENT

SURGERY

Polyp removal (polypectomy)

Colonic resection (colectomy)

- If multiple polyps associated with polyposis syndromes/polyps with high-grade dysplasia



Figure 29.1 The gross pathological appearance of a sessile colorectal polyp.

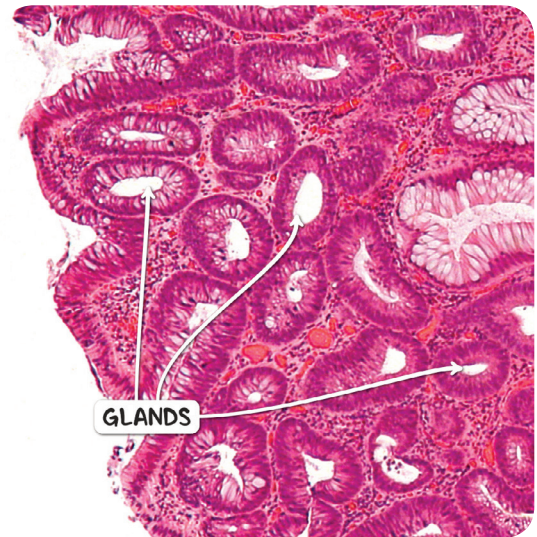


Figure 29.3 The histological appearance of a tubular adenoma composed of compact glands with variable levels of dysplasia.

FAMILIAL ADENOMATOUS POLYPOSIS (FAP)

osms.it/familial-adenomatous-polyposis

PATHOLOGY & CAUSES

- Inherited condition; hundreds/thousands adenomatous polyps in colon
- Autosomal dominant inheritance; 100% penetrance; de novo mutations may occur

TYPES

Classic FAP

- Most aggressive, frequent; > 100 polyps at diagnosis; early onset

Attenuated FAP (AFAP)

- < 100 polyps at diagnosis (oligopolyposis); later onset

Autosomal recessive FAP

CAUSES

- Germline mutation in APC gene (tumor suppressor) → prevention of apoptosis → cell overgrowth → polyps
- APC gene nonfunctional in FAP; slightly impaired in AFAP
- Autosomal recessive FAP
 - Mutations of MUTYH gene on chromosome 1

RISK FACTORS

- Family history

COMPLICATIONS

- Malignancy if untreated
- Extracolonic manifestations
 - Congenital hypertrophy of retinal pigment epithelium (CHRPE)
 - Fundic gland polyps: sessile polyps in stomach, usually not malignant
 - Duodenal adenomas: malignant potential

- Abdominal mesenchymal desmoid tumors: compress adjacent structures → obstruction/vascular impairment
- Other potential malignancies: thyroid, pancreas, brain (glioma), liver (hepatoblastoma)

SIGNS & SYMPTOMS

- Usually asymptomatic until malignancy
- Colonic manifestations
 - Palpable abdominal mass; hematochezia (rectal bleeding); pain (esp. abdomen); diarrhea

DIAGNOSIS

DIAGNOSTIC IMAGING

Endoscopy with biopsy

Colonoscopy, flexible sigmoidoscopy:

- Detection of ≥ 100 polyps; ~30 polyps, AFAP

Esophagogastroduodenoscopy (EGD)

- Gastric, duodenal adenomas

Barium enema (with double contrast)

- Filling defects

Abdominal CT scan

- Hyperdense outpouchings of colonic wall into lumen

LAB RESULTS

- Iron-deficiency anemia
- ↓ RBC, ↓ MCV
- ↓ ferritin, ↓ serum iron, ↓ transferrin saturation
- APC mutations

OTHER DIAGNOSTICS

Family history

- Cancers, gastrointestinal (GI) tract diseases

Digital rectal examination

- Palpable mass

Ophthalmic examination

- CHRPE

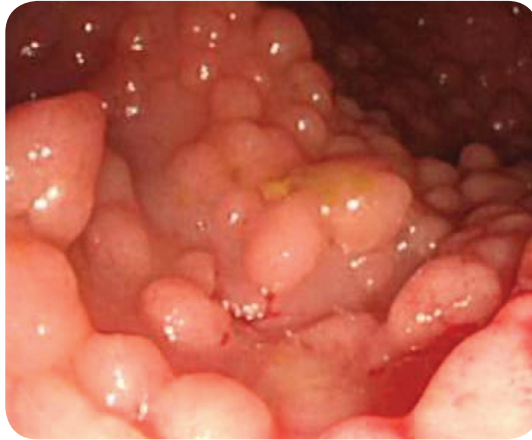


Figure 29.4 Endoscopic appearance of the colon in a case of familial adenomatous polyposis.

TREATMENT

MEDICATIONS

- Cyclooxygenase 2 inhibitors, other nonsteroidal anti-inflammatory drugs (NSAIDs)
- *Epidermal growth factor receptor inhibitor*: erlotinib
- Chemotherapy, if colon cancer

SURGERY

- Frequent endoscopic check-ups to detect onset of polyposis every 1–2 years
 - If polyps detected → surgical removal (colectomy; proctocolectomy)

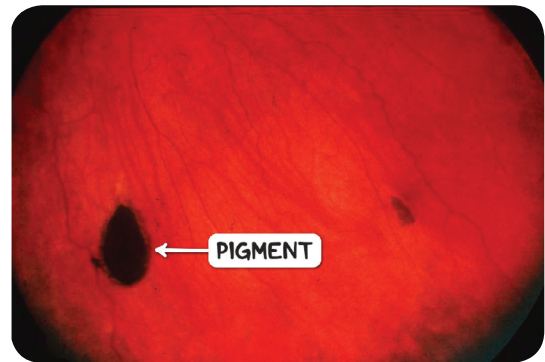


Figure 29.5 A retinal photograph demonstrating hypertrophy of the retinal pigment epithelium in a case of familial adenomatous polyposis.

GARDNER'S SYNDROME (GS)

osms.it/gardners-syndrome

PATHOLOGY & CAUSES

- Variant of FAP with prominent extracolonic manifestations
- Inherited condition; numerous adenomatous polyps in colon; extracolonic polyps, tumors
- Tumors outside colon
 - Fibromas, lipomas, epidermoid cysts, thyroid neoplasms, osteomas, desmoid
- Extracolonic polyps can arise in stomach, duodenum, spleen, kidneys, liver, mesentery, small bowel; CHRPE lesions

CAUSES

- APC, RAS, TP53 mutation; DCC deletion → furthers carcinogenesis
- Autosomal dominant inheritance

COMPLICATIONS

- Malignancy in colon, thyroid, liver, kidneys

SIGNS & SYMPTOMS

- Colonic manifestations
 - Rectal bleeding, diarrhea
- Extracolonic manifestations
 - Desmoid tumors (parietal bumps, bleeding)
 - Dental problems
 - Epidermoid cysts
 - Epigastric pain, bleeding, jaundice
 - Malnutrition → malaise, lethargy, fatigue

DIAGNOSIS

DIAGNOSTIC IMAGING

Endoscopy with biopsy

Colonoscopy, flexible sigmoidoscopy

- Direct visualization of adenomatous polyps in colon

Abdominal CT scan

- Hyperdense outpouchings of colonic wall into lumen

Head/dental X-ray

- Dental abnormalities

LAB RESULTS

- Iron-deficiency anemia
 - ↓ RBC, ↓ MCV
 - ↓ ferritin, ↓ serum iron, ↓ transferrin saturation
- Tumoral markers (e.g. carcinoembryonic antigen)
- APC, RAS, TP53 mutations; DCC deletion

OTHER DIAGNOSTICS

Physical examination

- Supernumerary impacted teeth
- Multiple jaw osteomas, odontomas

Digital rectal examination

- Palpable mass

Ophthalmic examination

- CHRPE

ECG

- Stomach, duodenum for polyps

TREATMENT

- No cure; palliative treatment

SURGERY

- Excision of tumors/polyps with wide (8mm) margin
- Colectomy

OTHER INTERVENTIONS

- Radiotherapy, if recurrent

JUVENILE POLYPOSIS SYNDROME

osms.it/juvenile-polypsis

PATHOLOGY & CAUSES

- Numerous **benign** (AKA juvenile) **polyps** along GI tract
- Majority non-neoplastic **hamartomas** **polyps**, in **colorectum**

CAUSES

- **BMPR1A**, **SMAD4** mutations
- Autosomal dominant inheritance; incomplete penetrance
- De novo mutations (25%)

COMPLICATIONS

- Increased risk of **colorectal/extracolonic adenocarcinoma**; intestinal obstruction

SIGNS & SYMPTOMS

- Hematochezia, anemia symptoms; abdominal pain; diarrhea/constipation; rectal prolapse

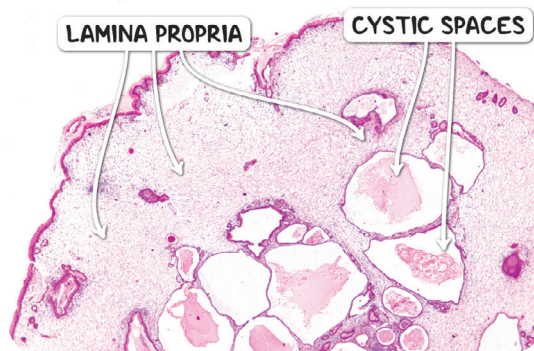


Figure 29.6 A juvenile retention polyp with abundant edematous stroma and dilated cystic spaces filled with mucin. The spaces are lined by cuboidal epithelium.

DIAGNOSIS

DIAGNOSTIC IMAGING

Endoscopic studies

- E.g. endoscopy, colonoscopy, sigmoidoscopy
- Criteria for diagnosis
 - > five juvenile polyps in colon/rectum
 - Multiple juvenile polyps in other areas of GI tract
 - Family history with any number of polyps
- Biopsy, cytology

LAB RESULTS

- Iron-deficiency anemia
 - ↓ RBC, ↓ MCV
 - ↓ ferritin, ↓ serum iron, ↓ transferrin saturation
- **BMPR1A**, **SMAD4** mutations

TREATMENT

SURGERY

- Polypectomy
- Surgical colectomy, proctocolectomy
 - Malignant, ulcerating polyps

PEUTZ–JEGHERS SYNDROME (PJS)

osms.it/peutz-jeghers

PATHOLOGY & CAUSES

- Inherited condition; **benign hamartomatous polyps**, in small bowel; also in colon, stomach
- Associated with **hyperpigmented** (melanin-containing) **macules on skin, mucosa**

CAUSES

- IV drug use
 - Increases likelihood of infective endocarditis
- Congenital bicuspid aortic valve
- Diabetes, high blood pressure, smoking

COMPLICATIONS

- Very high risk of **extracolonic malignant transformation**
 - Breast, ovarian, cervical, testicular, pancreatic, thyroid cancer
- Mild malignant potential of polyps

SIGNS & SYMPTOMS

- GI
 - Ulceration → GI bleeding (hematochezia/melena) → symptoms of anemia
 - Colicky abdominal pain
 - Intussusception → bowel obstruction, bowel infarction
 - Diarrhea, constipation
- Pigmented lesions around oral mucosa, nostrils, perianal area of extremities; fade after puberty

DIAGNOSIS

DIAGNOSTIC IMAGING

Endoscopy, colonoscopy, with biopsy

Capsule endoscopy

Abdominal CT scan

- Hyperdense outpouchings of colonic wall into lumen

LAB RESULTS

- Fecal occult blood test
- Iron-deficiency anemia
 - ↓ RBC, ↓ MCV
 - ↓ ferritin, ↓ serum iron, ↓ transferrin saturation
- Tumor markers
 - CEA, CA-19-9, CA-125
- STK11 (LKB1) mutations

OTHER DIAGNOSTICS

Diagnostic criteria

- One of following
 - ≥ two PJ polyps confirmed histologically
 - ≥ one PJ polyp with family history
 - PJS-associated mucocutaneous pigmentations

Digital rectal examination

- Palpable mass

TREATMENT

SURGERY

- Polypectomy

MEDICATIONS

- Cyclooxygenase 2 inhibitors (celecoxib)

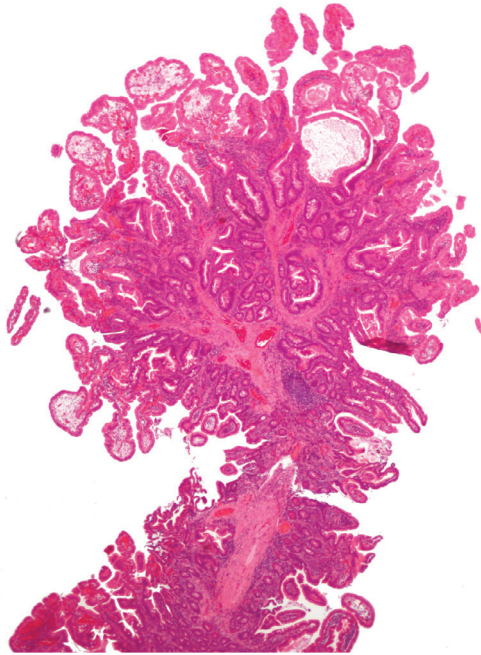


Figure 29.7 Histological appearance of a Peutz-Jegher's polyp.



Figure 29.8 Multiple melanotic macules on the skin and oral mucosa of a young boy with Peutz-Jegher's syndrome.